Quantifying the benefits and sustainability of paediatric precision medicine delivery

Precision medicine is at the frontier of modern medical science – but it is expensive and challenging to introduce into the health system. While we know it brings health benefits for many children, the long-term costs or effects on the health system are less clear.

Luminesce Alliance funding has supported research that will contribute to answering these questions.

In collaboration with the Australian Institute of Health Innovation (AIHI) at Macquarie University, implementation scientists and health economists studied ZERO to see how the research and healthcare staff who deliver the program interact and communicate.

The AIHI team studied the complex interactions involved in ZERO's implementation, using a mixture of research methods such as social network analysis (as shown above), rapid ethnography and interviews with clinicians and health staff.

By watching how their relationships change and develop over time, the researchers can advise on what needs to be improved to support the smooth rollout of the program and remove barriers.

"As far as we know, this is the first time anyone in precision medicine has put an implementation science lens of this kind alongside an intervention to change practice on the ground," says Prof Jeffrey Braithwaite, Director of AIHI.

"One of the biggest challenges with research is translating and transferring it into the health system to support better health outcomes," explains A/Prof Vanessa Tyrrell, Program Leader of ZERO, and Co-Head of Theme, Personalised Medicine, at CCI.

"If we want to embed the precision medicine model of care into the health system in the longer term, we need to understand what the barriers might be. We also need to understand the cost-effectiveness of precision medicine programs."

A/Prof Vanessa Tyrrell
Program Leader, Zero Childhood Cancer
Co-Head of Theme, Personalised Medicine
Children's Cancer Institute

The research team investigated the requirements of integrating precision medicine at scale in the paediatric health system.

"We were trying to understand what would be needed in terms of workforce planning and to identify potential bottlenecks or barriers we can address as we continue to expand the program," A/Prof Tyrrell says.

Issues highlighted by the researchers included a reliance on informal or invisible work by health professionals over and above their recognised roles, and the need for health workforce education in precision medicine.

Key to the successful implementation of ZERO is a whole of systems approach, workforce education, and the creation of extensive but previously non-existent connections and networks. However, the analysis also found a heavy reliance on informal roles and key individuals, an invisible workforce challenge to overcome.

"Our study highlighted that the whole system should be considered in all of its complexity rather than reducing it into constituent parts," Prof Braithwaite says.

Cost analysis

Previous research into the effectiveness and cost-effectiveness of genomic sequencing for rare diseases has shown significant cost savings. However, there has been no similar work done in the paediatric cancer context.

A health economics project has aimed to address this, done as a collaboration between ZERO and the Centre for Economic Impacts of Genomic Medicine (GenIMPACT) at Macquarie University. The researchers performed an economic analysis of ZERO, the first analysis of its kind specifically microcosting the comprehensive approach ZERO takes to precision cancer medicine in a paediatric cancer setting.

The initial findings indicate that costs were lower than previous studies, that this is likely due to continued downward trend in consumables costs and improved automation of computational and analysis pipelines, and that costs will continue to trend down over the next three years.

The findings of this soon-to-be published research and future studies will guide funders and policy makers in making better informed decisions when considering how to implement precision medicine in the care of children with cancer in the future.

Re-writing medical history and transforming lives

A multidisciplinary collaboration has radically shifted the model of care for the detection and treatment of a devastating genetic motor neurone disease, transforming the lives of families.

Spinal muscular atrophy (SMA) affects one in 10,000 births and was once the leading genetic cause of infant death. In its most common and most severe form, it quickly paralyses babies, who survive on average nine to 10 months. While their brains remain unaffected, they lose the ability to move, feed and ultimately breathe.

It is now possible to screen newborn babies for the disease and access life-saving gene therapy for those likely to develop severe SMA.

The breakthrough is the result of groundbreaking medical research and clinical trials by specialists from across SCHN and the Universities of Sydney and NSW Sydney. These teams worked together to prove the efficacy of screening and health economic benefits of a novel gene therapy developed and manufactured by an international pharmaceutical company.

"The success was achieved through many people coming together and the Luminesce Alliance helped to enable that," says A/Prof Michelle Farrar, paediatric neurologist at Sydney Children's Hospital and UNSW Sydney.

"We know that early identification is vital in the treatment of SMA and that is what the newborn screening program has allowed us to do. It has radically shifted our model of care and we are now in a position where we can rewrite the history of SMA."